



## hepatic veno-occlusive disease with immunodeficiency

Hepatic veno-occlusive disease with immunodeficiency (also called VODI) is a hereditary disorder of the liver and immune system. Its signs and symptoms appear after the first few months of life.

Hepatic veno-occlusive disease is a condition that blocks (occludes) small veins in the liver, disrupting blood flow in this organ. This condition can lead to enlargement of the liver (hepatomegaly), a buildup of scar tissue (hepatic fibrosis), and liver failure.

Children with VODI are prone to recurrent infections caused by certain bacteria, viruses, and fungi. The organisms that cause infection in people with this disorder are described as opportunistic because they ordinarily do not cause illness in healthy people. These infections are usually serious and may be life-threatening. In most people with VODI, infections occur before hepatic veno-occlusive disease becomes evident.

Many people with VODI live only into childhood, although some affected individuals have lived to early adulthood.

### Frequency

VODI appears to be a rare disorder; approximately 20 affected families have been reported worldwide. Most people diagnosed with the condition have been of Lebanese ancestry. However, the disorder has also been identified in several individuals with other backgrounds in the United States and Italy.

### Genetic Changes

VODI results from mutations in the *SP110* gene. This gene provides instructions for making a protein called SP110 nuclear body protein, which is involved in the normal function of the immune system. This protein likely helps regulate the activity of genes needed for the body's immune response to foreign invaders (such as viruses and bacteria).

Mutations in the *SP110* gene prevent cells from making functional SP110 nuclear body protein, which impairs the immune system's ability to fight off infections. It is unclear how a lack of this protein affects blood flow in the liver.

### Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal

recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

### **Other Names for This Condition**

- familial veno-occlusive disease with immunodeficiency
- hepatic venoocclusive disease with immunodeficiency
- veno-occlusive disease and immunodeficiency syndrome
- VODI

### **Diagnosis & Management**

#### Genetic Testing

- Genetic Testing Registry: Hepatic venoocclusive disease with immunodeficiency  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1856128/>

#### Other Diagnosis and Management Resources

- GeneReview: Hepatic Veno-Occlusive Disease with Immunodeficiency  
<https://www.ncbi.nlm.nih.gov/books/NBK1271>

#### General Information from MedlinePlus

- Diagnostic Tests  
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy  
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling  
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care  
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation  
<https://medlineplus.gov/surgeryandrehabilitation.html>

### **Additional Information & Resources**

#### MedlinePlus

- Health Topic: Immune System and Disorders  
<https://medlineplus.gov/immunesystemanddisorders.html>
- Health Topic: Liver Diseases  
<https://medlineplus.gov/liverdiseases.html>

### Genetic and Rare Diseases Information Center

- Hepatic venoocclusive disease with immunodeficiency  
<https://rarediseases.info.nih.gov/diseases/10083/hepatic-venoocclusive-disease-with-immunodeficiency>

### Additional NIH Resources

- National Institute of Allergy and Infectious Diseases: Primary Immune Deficiency Diseases  
<https://www.niaid.nih.gov/diseases-conditions/primary-immune-deficiency-diseases-pids>

### Educational Resources

- Disease InfoSearch: Hepatic venoocclusive disease with immunodeficiency  
<http://www.diseaseinfosearch.org/Hepatic+venoocclusive+disease+with+immunodeficiency/3331>
- MalaCards: hepatic venoocclusive disease with immunodeficiency  
[http://www.malacards.org/card/hepatic\\_venoocclusive\\_disease\\_with\\_immunodeficiency](http://www.malacards.org/card/hepatic_venoocclusive_disease_with_immunodeficiency)
- Merck Manual Home Edition for Patients and Caregivers: Veno-Occlusive Disease of the Liver  
<http://www.merckmanuals.com/home/liver-and-gallbladder-disorders/blood-vessel-disorders-of-the-liver/veno-occlusive-disease-of-the-liver>
- Orphanet: Hepatic veno-occlusive disease-immunodeficiency syndrome  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=79124](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=79124)

### Patient Support and Advocacy Resources

- American Liver Foundation  
<http://www.liverfoundation.org/>
- Children's Liver Disease Foundation (UK)  
<http://www.childliverdisease.org/>
- Immune Deficiency Foundation  
<http://primaryimmune.org/>
- International Patient Organisation for Primary Immunodeficiencies  
<http://www.ipopi.org/>
- Jeffrey Modell Foundation  
<http://www.info4pi.org/>
- Resource list from the University of Kansas Medical Center: Immune Deficiency Conditions  
<http://www.kumc.edu/gec/support/immune.html>

## GeneReviews

- Hepatic Veno-Occlusive Disease with Immunodeficiency  
<https://www.ncbi.nlm.nih.gov/books/NBK1271>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28hepatic+veno-occlusive+disease%5BTIAB%5D%29+AND+%28immunodeficiency%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

## OMIM

- HEPATIC VENOOCCLUSIVE DISEASE WITH IMMUNODEFICIENCY  
<http://omim.org/entry/235550>

## **Sources for This Summary**

- Cliffe ST, Wong M, Taylor PJ, Ruga E, Wilcken B, Lindeman R, Buckley MF, Roscioli T. The first prenatal diagnosis for veno-occlusive disease and immunodeficiency syndrome, an autosomal recessive condition associated with mutations in SP110. *Prenat Diagn.* 2007 Jul;27(7):674-6.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/17510920>
- GeneReview: Hepatic Veno-Occlusive Disease with Immunodeficiency  
<https://www.ncbi.nlm.nih.gov/books/NBK1271>
- Roscioli T, Cliffe ST, Bloch DB, Bell CG, Mullan G, Taylor PJ, Sarris M, Wang J, Donald JA, Kirk EP, Ziegler JB, Salzer U, McDonald GB, Wong M, Lindeman R, Buckley MF. Mutations in the gene encoding the PML nuclear body protein Sp110 are associated with immunodeficiency and hepatic veno-occlusive disease. *Nat Genet.* 2006 Jun;38(6):620-2. Epub 2006 Apr 30.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/16648851>

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